Indian J Med Res 152 (Supplement), November 2020, pp 67-68
DOI: 10.4103/ijmr.IJMR_2037_19

Quick Response Code:

A case of type-1 Gaucher disease

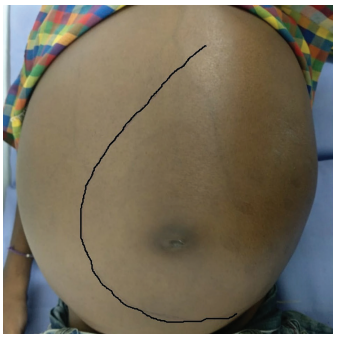

Fig. 1. Surface marking (black line) of the spleen. According to Hackett spleen classification, this is Grade 5 splenomegaly (spleen expands up to pubic symphysis).

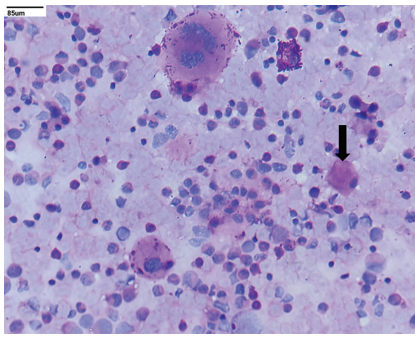

Fig. 2. Bone marrow biopsy showing Gaucher cell (black arrow) on periodic acid–Schiff staining (×40).

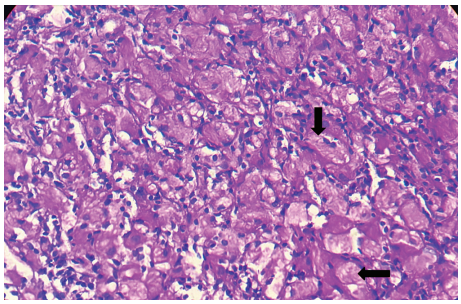

Fig. 3. Spleen biopsy showing abundant Gaucher cells (black arrows) with eccentric nuclei and crumpled tissue paper appearance of cytoplasm on periodic acid–Schiff staining (×40).

An 11 yr old male child[†] was presented to the department of Paediatrics, G.K. General Hospital, Bhuj, Gujarat, India, in July 2018, with chief complaint of abdominal distension for the last 12 months. On examination, severe pallor, gross abdominal distension and massively enlarged spleen were present (Fig. 1). Developmental quotient was normal (88%). Weight for age and height for age were <−3 standard deviation, and body mass index was 15.9. Pancytopenia was present, and on bone marrow examination, Gaucher cells were seen with periodic acid-Schiff (PAS) stain (Fig. 2). On spleen biopsy, abundant Gaucher cells were seen with peripherally placed nuclei (Fig. 3). Blood acid beta-glucocerebrosidase enzyme level was <10 per cent of normal.

[†]Consent to publish clinical information and images obtained from patient's parent.

Based on clinical presentation, histopathological findings and enzymatic assessment, a final diagnosis of type-1 Gaucher disease was made. Supportive treatment was given initially, and enzyme replacement therapy (ERT) was advised. However, even after two months ERT was not initiated and the patient was lost to follow up thereafter.

Gaucher disease should always be considered in the differential diagnosis of huge splenomegaly. ERT is the standard treatment of Gaucher disease and should be started as soon as diagnosis is confirmed.

***Acknowledgment*:** Authors acknowledge Dr Dhaneshwar Lanjewar from the department of Pathology, Gujarat Adani Institute of Medical Sciences, Bhuj, for histopathological examination and assisting in diagnosis.

***Conflicts of Interest*:** None.

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Received November 10, 2019